APPENDIX B2: CLEAN COPY OF SPECIFICATION

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FIGS. 1A-1B are a DNA sequence of the human FKHL7 gene including the 5' and 3' untranslated regions (UTRs) (SEQ ID No. 1). The 1659 base pair open reading frame is provided herein as SEQ ID NO. 3 and the (SEQ ID NO. 3) 553 amino acid human FKHL7 protein is provided herein as SEQ ID No. 2. The forkhead region of the protein is indicated by underline.

Paragraph bridging pages 7-8:

FIG. 2 shows an amino acid comparison of the forkhead domains of different members of the FKHL-family of genes. The locations of the three alpha helices and the two wing domains are shown (Clark, K.L. et al., Nature 364:412:420 (1993)). The *Drosophila* forkhead gene sequence is shown above that for *FKHL7*, while the positions of the three missense mutations are shown below *FKHL7*. Translation of the 11 base pair deletion (bp del) mutation results in total loss of the forkhead domain. The other FKHL family members are shown below *FKHL7* for comparison. For *FKHL10*, only partial sequence is available for forkhead domain. The last sequence shown is that for the distantly related *FKHR* which has been mapped to 13Q14 near the *RIEG2* locus.

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FIG. 3 provides the identity and location of Expressed Sequence Tags (ESTs) that map to regions of the human FKHL7 gene.

Paragraph bridging pages 91-92:

An 11 bp deletion upstream of the *FKHL7* forkhead domain was identified in two brothers diagnosed with different anterior segment defects (RA and IH). Both brothers had

glaucoma, and neither had the extra-ocular manifestations of Reiger syndrome (RS). Their father was found to have isolated posterior embryotoxon (PE), suggesting that the disease was inherited through him as an autosomal dominant. He was also found to carry the deletion. A second mutation was found in a proband and her mother who were both diagnosed with classic RA and glaucoma. This mutation, a C to T transition within the forkhead domain causes a change from a serine to a leucine (SER131Leu). A third mutation, a C to G transversion within the forkhead domain, was found in a proband with severe Axenfeld anomaly and glaucoma. This change results in the replacement of isoleucine with methionine (Ile126MET) and is also found in the father who was diagnosed with AA. Finally, a T to C transition was found in a proband of an extended family with a spectrum of anterior segment defects. This change results in the replacement of phenylalanine with serine (Phe112Ser) within the forkhead domain. Three of the mutations were not found in 128 unrelated normal individuals from an ethnically similar control population (Caucasian). The fourth mutation (Phe112Ser) was only detected by direct sequencing of PCR products from patient genomic DNA. This mutation was found to segregate with the disease in an extended pedigree and was not present in an additional 12 Caucasian individuals by sequence analysis.